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Reconstruction of an Upper Lip and the Coloboma in the Nasal Ala Accompanying with Freeman-Sheldon Syndrome

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In 1938 FREEMAN and SHELDON described two cases with the characters of face, hand and feet. They gave the name as craniocarpo-tarsal dystrophy. After this publication, OTTO (1953), WALKER (1960) and KULZ (1961) described similar cases. BURIAN (1963), without knowledge of the previous description of FREEMAN and SHELDON, described three cases and titled as the "whistling face" characteristic of a compound cranio-facio-corporal syndrome. CERVENKA and GORLIN (1969 & 1970) described the results of measurement in oral intercommissural distance between white Czechoslovakian children and American children to establish norms. pitanguy in 1969 depicted a chart of their generations with this syndrome.

Report of a case

The patient was born March 12, 1974. His mother did not take anykind of drugs during her pregnancy. The family history was negative for similar anomalies. There were no history of parental consanguinity. The mother and father were 21 and 26 years old at the time of birth of the patient respectively. Birth at sacral position, the baby's weight at birth was 3,040 g. Because oral intercommissural distance was very narrow, the baby could not suck up the breast-fed. So, he was reard by a artificial feeding. Various anomalies of the baby was pointed out in post-parturition by the obstetrician as following. Torticolli muscularis, club foot, congenital dislocation of the hip and face anomalies. The baby was made admission to orthopaedic surgery clinic to treat for torticolli, dislocation of the hip and club foot.

The results of routine laboratory examination including blood biochemistry, urine and chromatin were normal. M. orbicularis oris showed the lower spike by EMG.

X-ray finding: In comparison with the head broaden to the frontal plane, the lower part of the face is narrow. The skull revealed a steep anterior cerebral fossa to cranio-facial disproportion (Fig. 1).

In the measurement on Down's method, following results were obtained: a facial angle 78°, angle of maxillary convexity 10°, A-B plane 7°, Frankfort mandibular plane angle 43° and Y axial angle 90° Index finger on the left hand reveals ulnar deviation (Fig. 2).

Key words: Broader skull, Microstomia, Coloboma, Clinodactly, Club foot.
索引語：頭蓋拡大，小口症，鼻翼欠損，屈指，内反足.
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Fig. 1 a.

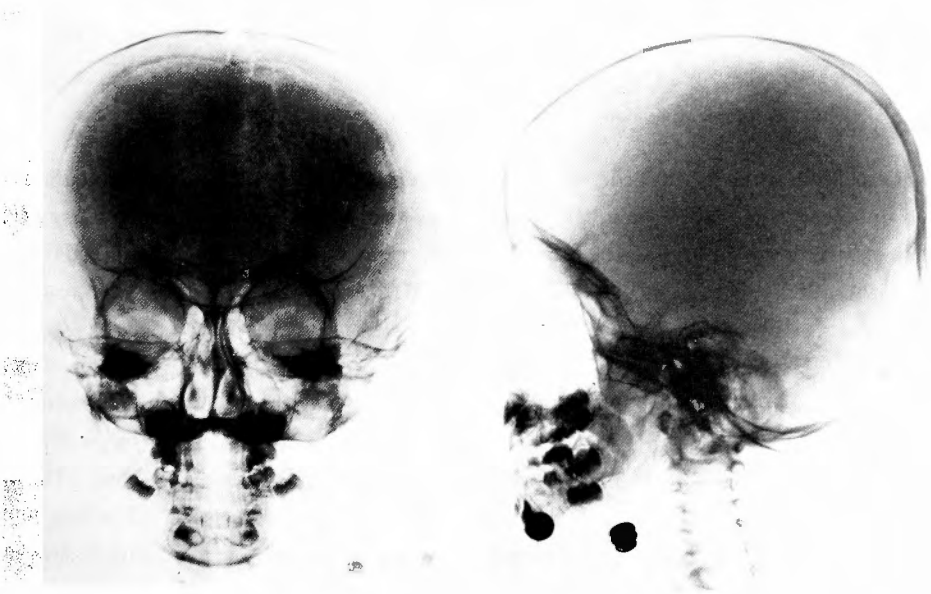

Fig. 1 b.

Fig. 1. a) Coloboma of the nasal ala and microstomia
b) X-ray shows a steep anterior cerebral fossa and broader skull

Treatment: Though the blepharophimosis, microstomia and coloboma of the ala became the object of surgical procedures, Author has done the reconstruction of the microstomia, depressive philtrum and coloboma of the ala. Atempt of the oral opening was made by Kazanjian's method and 1.3 cm incision was made bilaterally. But the contracture was not able to release (Fig. 3).

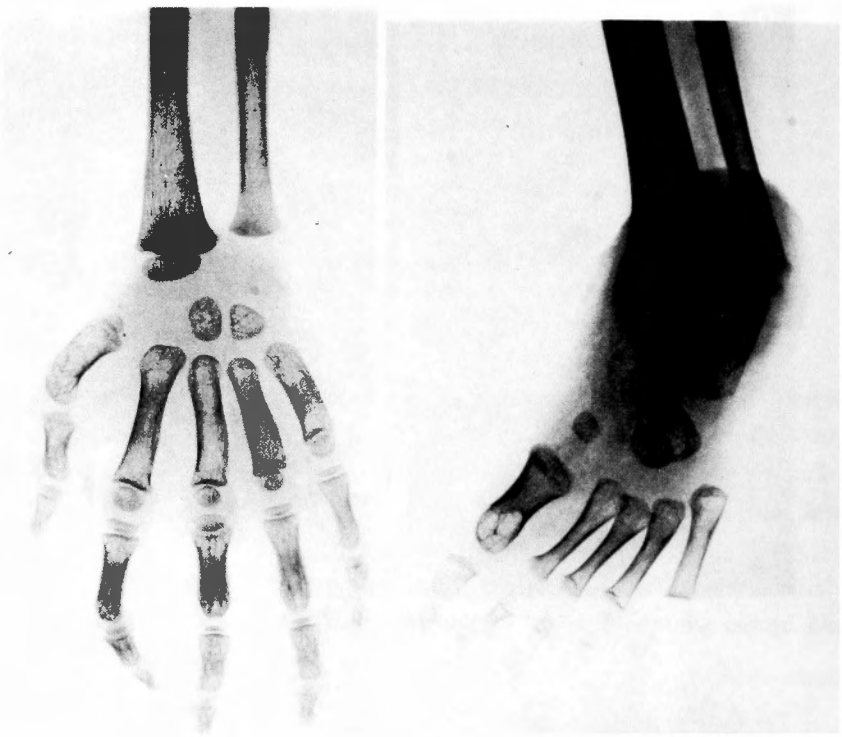

Fig. 2. Photograph shows ulnar deviation of index finger at PIP joint and club foot.

Quadral incision at the inner border of the coloboma to repair the ala was made, quadral skin flap turned up toward the alar rim, which was implanted with a piece of the ear cartilage (Fig. 4). The mucosal V incision to repair the thin philtrum dimple was done from upper labial frenulum to vermillion border. The muscles at the philtrum area had thinner layer and the greater part of M. orbicularis oris revealed massive form at the philtrum ridge on the both side (Fig. 5). These massive muscles are undermined, pulled from each side and sutured at the midportion of the upper lip. Finally, turning of mucosal flap was sutured to make V form to the original position.

Pathological findings: Biopsy of M. orbicularis oris, obtained during surgical procedure, were stained with hematoxylineosin, toluidine blue, Masson, and PAS-alcian blue. The muscles revealed irregular range, split, vacuous, and obscure straiated structures. Position, form and ranged of the muscular nucleus were irregular. Lot of loose fibrous connective tissues between muscle bundle were found (Fig. 6).

Discussion

Reports in regarding Freeman-Sheldon syndrome are less than 45 cases. Moreover, the present author could not found the descriptions of surgical treatment except BURIAN (1963) and WEINSTEIN (1969). BURIAN make paralled incision to the border of the ala and he prolonged the inner lining by an inverted V Y plasty. Other, Weinstein made the enlargement of oral

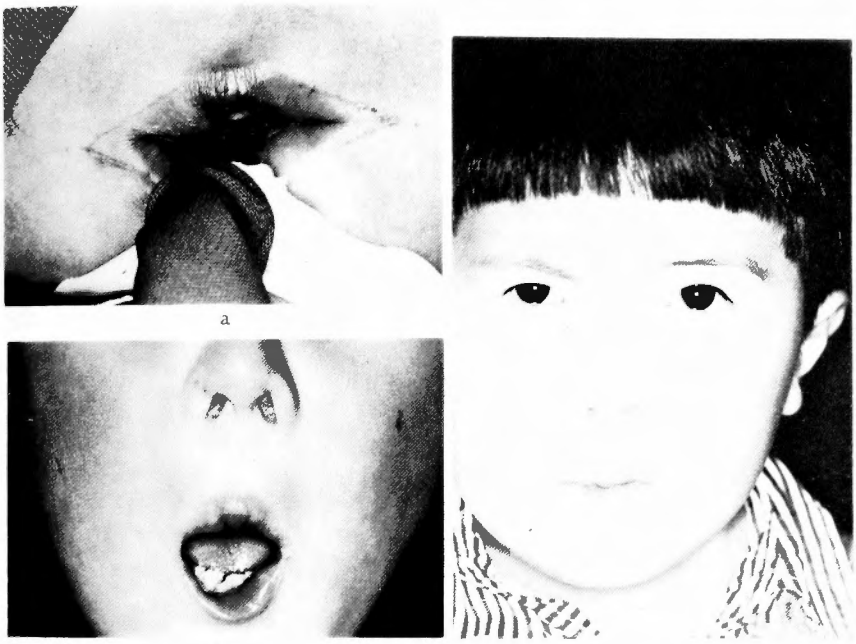

Fig. 3. a) Oral openings design
b) Opening profile after two years shows remaining of contracture at oral angle
c) Deep setting eye on the left side and deformity of nasal ala after implantation of ear cartilage

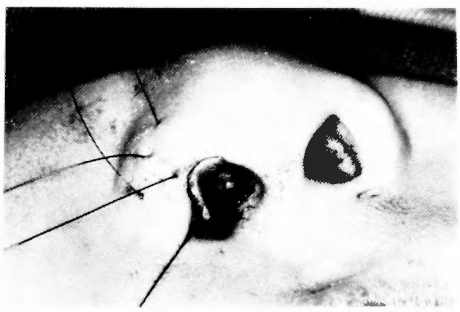

Fig. 4. Implantation with a piece of the ear cartilage

opening by bilateral Y-shaped incisions and triangular flaps laid to construct new commissures. BURIAN (1963), GORLIN (1974) and SAUK (1974) reported the descriptions on the pathologic feature that the muscle fiber and cells presented degenerative changes. Otherwise, though they described also in regarding to the results of the electromyographic examination on the several facial muscles, that results confirmed only lower spikes of M. buccinator. As they have ever reported, present case also had not revealed abnormal EMG findings.

Aetiology of this syndrome is unknown. PITANGUY (1969) reported six cases, five of six

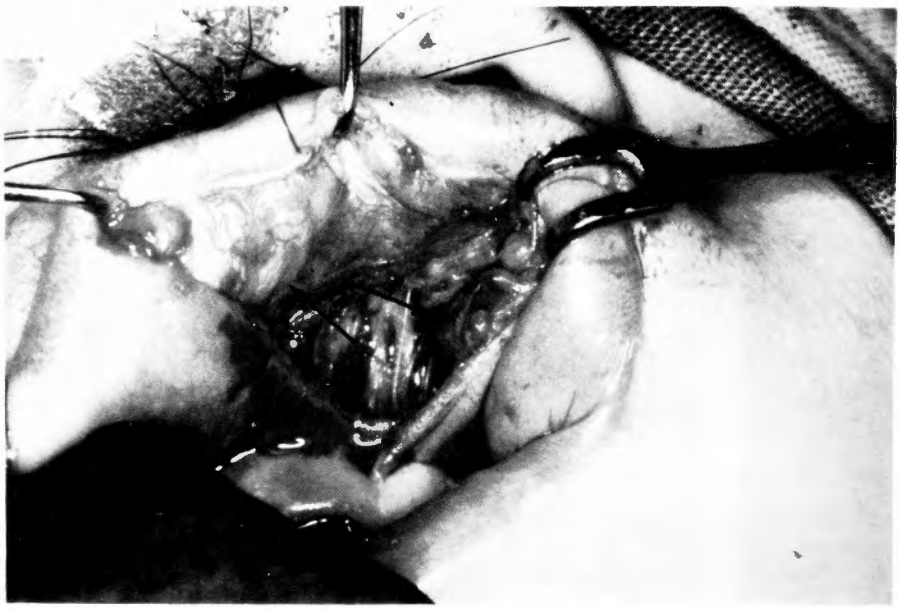

Fig. 5. Massive formation of M. orbicularis oris at the philtrum ridge on the both sides

cases were members of the same family and of three consecutive generations. FRASER (1970) reported the cases of the father and his son. GROSS-KIESELSTEIN (1971), PFEIFFER (1972), and BARTA (1973) reported each cases of the mother and her daughter. WALKER (1960) presented a pedigree of three consecutive generations. PITANGUY described that appearance of the syndrome might be affected by a familiar gene of dominant and irregular character. WEIN-

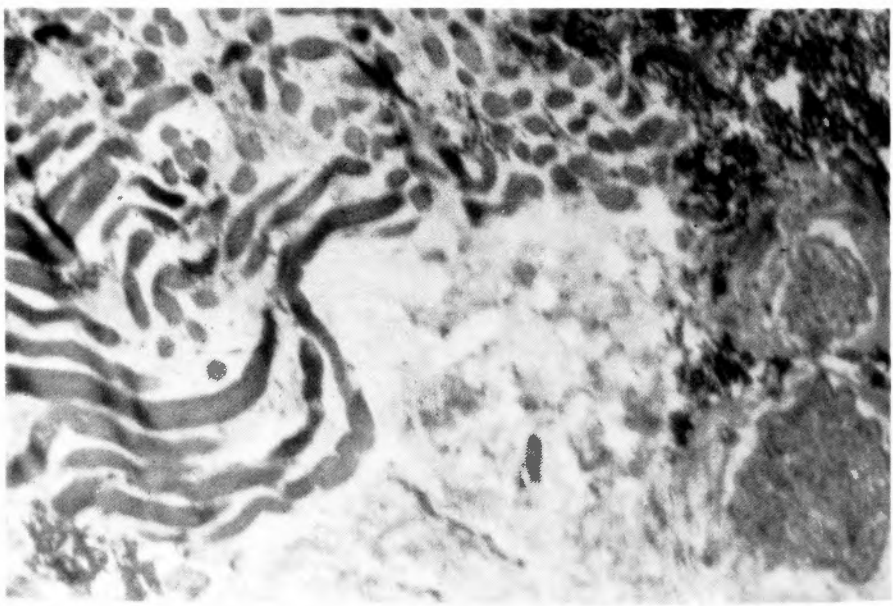

Fig. 6. The muscle fibers are irregular range, split, vacuous, and obscure straiated structures

STEIN, GORLIN and GROSS-KIESELSTEIN suggests an autosomal dominant inheritance. Other, FRASER offered the sporadic cases which would have a very lower risk of being affected, offspring would have a 50 : 50 chance. As FRASER offered the mutations, under the limitation of the present case, it would like to support his hypothesis because nobody was appearance of some anomalies in the family. HASHEMI (1973) indicated a case following the malformation of the kidney, GORLIN (1974) reported a case accompanying pterygium colli. From view points of the embryology and its genetic, it will be suppose that the patients of this syndrome has so many risks accompanying anomalies.

The present author could not yet search out a case report that associated with congenital luxation of the hip following this syndrome.

Summary

1) A case with characteristic face and associated with ulnar deviation of index finger, club foot congenital dislocation of hip was presented.
2) Coloboma of the nasal ala was repaired by the implantation of piece of the ear cartilage, small mouth was enlarged bilaterally by Kazanijan's method and the disruptions of muscle in midportion were sutured at the correct position. But, grafted ear cartilage was completely absorbed after 2 years.
3) Biopsy of M. orbicularis oris showed degenerative findings of muscles.

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和文抄録

Freeman-Sheldon 症候群の口唇および鼻翼再建例

岩手医科大学形成外科

奈 良 卓

Freeman-Sheldon 症候群は非常に稀な先天異常であり，頭蓋，顔面，四肢などの異常を伴うのが特徴で，この異常のうち小口症および鼻翼部分欠損に対し Kazanjian 法による口角形成を行い開口の改善を図りまた，鼻翼には耳介軟骨移植を行った．組織的に口輪筋線維束の走行異常，筋線維の萎縮変性，結合織の増殖を認めた．